

Osteomas in a Patient with Familial Adenomatous Polyposis

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Rec date: July 18, 2017; Acc date: August 02, 2017; Pub date: August 07, 2017

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Abstract

This case report aimed to characterize dento-osseous anomalies in Familial Adenomatous Polyposis (FAP) in a Brazilian patient. Furthermore, FAP was investigated for possible causative. This case report showed the importance of dento-osseous knowledge related to FAP. Early dento-osseous anomalies diagnosis revealed the need to follow-up FAP family members from childhood and was essential for subsequent clinical or genetic FAP diagnosis. The authors think that this work is important as it provides highlights about the role of the dentist in the early diagnosis of FAP, a disease that predisposes colorectal cancer.

Keywords: Inherited diseases; Chromosome 5q21; Colorectal polyps; Dento-osseous lesions; Bone anomalies

prevalence of FAP has been estimated to be approximately 1 in 10,000 live births [4,5].

Introduction

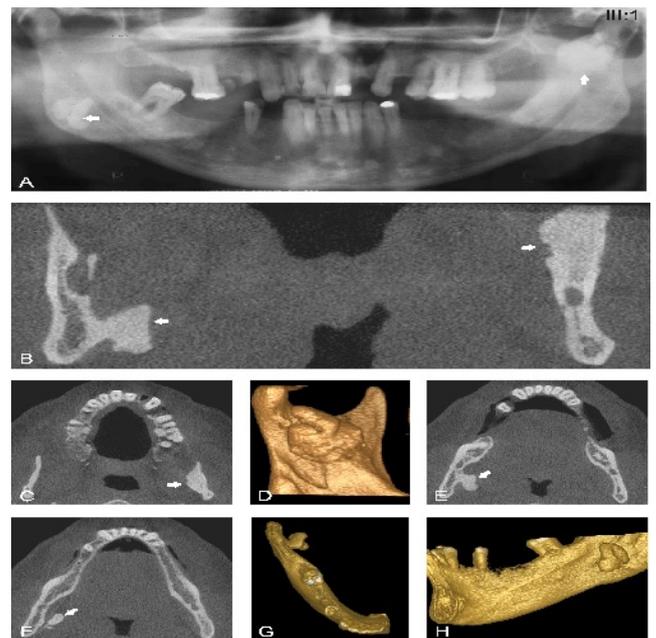
A 41-year-old male patient was referred to Oral Care Center for Inherited Diseases, University of Brasilia, Brasilia, Brazil, with a presumptive diagnosis of Temporomandibular joint (TMJ) disorders. Intraoral examination revealed pain and limited mouth opening. The medical history and general physical condition were obtained and the patient reported recent diagnosis of gastrointestinal polyps.

Case Report

Dental panoramic radiographs revealed two radiopaque masses suggesting osteomas, one on the right mandibular angle region, and the other on the left side of the mandible, near the notch region (Figure 1A, white arrows). Furthermore, an increased diffuse radiopacity was observed in both jaw bones (Figure 1A). Based on the clinical and imaging findings, it could be concluded that the osseous changes were related to Familial Adenomatous Polyposis, rejecting the previous diagnosis of TMJ disorder. The bone anomalies and their relationship with the adjacent anatomic structures were more precisely assessed on cone-beam computed tomography scans (Figures 1B-1H). The patient was referred from Division of Coloproctology of the University Hospital of Brasilia (Brasilia, Brazil). A colonoscopy was performed following genetic analysis. A next-generation sequencing panel was made for the APC gene (Ion Ampliseq Custom Panels, Thermo Fisher) that confirmed the diagnosis of familial adenomatous polyposis.

Discussion

Familial adenomatous polyposis (FAP) (OMIM #175100) is an autosomal dominant inherited CRC syndrome caused by a germline mutation of the adenomatous polyposis coli (APC) gene located at chromosome 5q21 [1,2]. APC is a tumor suppressor gene expressed in a variety of human tissues including colorectal epithelium, and their inactivation constitutes the initial step in the development of CRC [3]. The majority of patients with FAP have a family history. The



Figures 1A-1H: A: Dental panoramic radiograph showing two radiopaque masses suggesting osteomas, one on the right mandibular angle region, and the other at the left side, near the notch region (White arrows). An increased diffuse radiopacity was also observed in both jaw bones. B to H: Cone-beam computed tomography images revealing bilateral osteomas and their relationship with anatomic structures. B: Coronal reformed image. C: Axial scan. D: 3D image, superior view. E: Axial scan. F: Axial scan. G: 3D image, axial view. H: 3D image, lateral view.

In addition to colorectal manifestations, patients with FAP can develop a variety of extra colonic alterations such as other types of

cancers, gastric and duodenal polyps, desmoids tumors, hypertrophy of the retinal pigment epithelium, epidermal cysts, and oral manifestations. Oral manifestations can emerge before the development of the colorectal polyps and are reported to be present from 58% to 100% of affected individuals [6-13]. The oral manifestations of FAP include osseous jaw lesions, odontomas, supernumerary teeth, unerupted teeth, and oral mucosa alteration. A recent meta-analysis showed that the frequency of osseous jaw and dental anomalies in affected individuals were 65.35% and 30.48%, respectively [14]. There has been growing interest in characterizing these alterations because they may precede CRC and may be used as an early disease diagnostic marker [15-20].

Conclusion

In conclusion, this case report showed the importance of studying dento-osseous lesions related to FAP. The early diagnosis of such bone anomalies reinforces the need to follow-up FAP family members from childhood and may be essential for subsequent clinical or genetic FAP diagnosis.

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